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EXPERIENCE WITH SOME UNUSUAL DISEASES OCCURRING IN FAMILIES

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The familial incidence of many disease and anomalies is well known. The pattern of heredity in some of these, such as hemophilia, has been well described. From time to time, attention has been called to the familial incidence of some diseases not usually regarded as hereditary. Sarcoidosis has been described in identical twins. Recently, Buerger's disease has been found in a brother and sister, although many instances have been recorded in which brothers had the disease. A colleague of mine recently observed polycythemia vera in identical twins, 7 years of age. The purpose of this paper is not World War II, involving certain unusual and curious disorders occurring in several families. The familial aspects of these have not been emphasized.

DISSEMINATED LUPUS ERYTHEMATOSIS

The familial susceptibility to disseminated lupus erythematosis has received some attention and has been described in two siblings. I will discuss briefly a family of six children, three of whom have died of the disease. The first case died at the age of ten years. She exhibited a typical skin eruption, fever, pallor, purpura, stomatitis and died in spite of supportive measures.

The second case was 20 years old when she died and had been ill for only 9 months. There was a marked anemia, thrombocytopenia and relative leukopenia, prolonged fever, splenomegaly and purpura but did not have the usual skin eruption. Autopsy, however, revealed the usual findings of disseminated lupus.

The third case in this family was 19 years old when she died and the total duration of her illness was only five months. The onset of her illness was characterized by mild joint discomfort and a sore mouth and she soon developed anemia, purpura, splenomegaly and a typical skin eruption over her face, palms and soles. She had a positive Wasserman from the beginning but there was no evidence of syphilis. Here again, autopsy confirmed the diagnosis of disseminated lupus. I should mention that all these cases died before the days of adequate hormone therapy.

The rest of this family has been examined. Neither the father nor the mother have ever shown evidence of the disease and nothing further was known of their ancestors. One sister proved to have a mild, hypochromic anemia and splenomegaly. Her platelet count and bone marrow studies were not unusual, although she did have a mild purpura. The youngest sister, age 14, was shown to have hypochromic anemia, splenomegaly and a reticulocytosis of 4%. Neither of these two girls have subsequently developed sufficient clinical evidence to make a diagnosis. However, these findings suggest that the final chapter in this family's susceptibility to the disease may not as yet be written. The brother presented a normal clinical picture.

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PORPHYRIA

It is well known that porphyria may occur in acquired or familial form. The clinical picture of the familial type has been well described and will not be mentioned. Early in 1948 a young woman was observed to have acute porphyria. She had none of the clinical stigma of the congenital type. She was seen post-operative because she was passing dark urine. She had been operated on because of acute abdominal pain but no pathology was found. She had taken various medications for dysmenorrhea prior to the surgery. The dark urine was shown to contain excessive amounts of porphyrins and she ran a typical course of acute porphyria, including extensive peripheral nerve damage. She made a complete recovery but since that time has had two exacerbations of her disease and, during the interval, has consistently shown abnormal amounts of porphyrins in the urine.

It was learned that her brother had a similar illness. He exhibited no stigma of congenital porphyria. His urine contained abnormal amounts of porphyrins each time that it was examined. His history certainly suggested that he had experienced an acute episode of porphyria. I mention these merely to show that what may appear to be acquired porphyria may be the congenital disease in exacerbation.

MACROCYTIC ANEMIA

Perhaps more interesting than the experiences just related is the observation of three sisters with a peculiar variety of anemia. This anemia has been characterized by chronicity, macrocytosis, an increased number of megaloblasts in the bone marrow, episodes of purpura and thrombocytopenia, a normal hemoglobin content of the cells, the presence of free hydrochloric acid in the stomach, and the failure to show significant response to substances providing the erythrocyte maturation factor, iron or thyroid extract. None have histories to suggest steatorrhea or episodes of hemolysis, and two have failed to respond to splenectomy. None have shown evidence of liver disease. Representative blood counts show some of the similarities. They had all been treated for a period of time by different physicians for the anemia without success. Interestingly enough, they have felt well and carried on normal activities in spite of the degree of anemia. One sister and one brother of these patients have normal counts, as does the mother who is the only living parent.

WOLFF-PARKINSON-WHITE SYNDROME

In 1949 an electrocardiogram was taken on a high school student because of a pulmonic systolic murmur, Grade I. She showed the typical findings of a short PR interval and a prolonged QRS complex characteristic of Wolff-Parkinson-White syndrome. In view of the experiences just related, arrangements were made to take electrocardiograms of her six siblings and of her mother and father. Two of the sisters showed almost identical electrocardiographic patterns. Similar cardiac silhouettes existed in these, as did a soft pulmonic systolic murmur. To my knowledge this is the first known incidence of Wolff-Parkinson-White syndrome occurring in siblings.

Later, the only brother in the family was apparently well, clinically, and did not have a heart murmur or electrocardiographic evidence of the W.P.W. syndrome but dropped dead in school. It is known that sudden death is not uncommon in this anomaly and it is known that it may exist only at times during one's life. He may have actually been the fourth case of the W.P.W. syndrome in this family.

POST-MENOPAUSAL OSTEOPOROSIS

In 1947 a 75-year-old woman presented herself complaining of acute backache. She had experienced chronic backache for years. There was a girdle-like pain over the lower abdomen and x-rays of the spine revealed diffuse osteoporosis with wedge-shaped fractures of several dorsal vertebra. Review of the previous x-rays taken in 1938 showed that the osteoporosis had existed at that time. Appropriate blood and urine chemistry studies were done to exclude osteoporosis other than the post-menopausal type. These were all negative. She made a symptomatic response following estrogen-androgen therapy.

Since that time, two of her three sisters have been observed. One of these sisters had back pain for years and studies established a similar diagnosis of her five, wedge-shaped fractures, noticed in the dorsal and lumbar spine, which were assumed to be spontaneous because there was no history of injury. Her back pain responded to hormone therapy and she was restored to an active life.

The second sister was seen because of a traumatic fracture and there was radiographic evidence of extensive osteoporosis. The third sister, who was not observed, died at the age of 68 in 1934 as a result of a transection of the spinal cord following a pathological fracture of the lumbar vertebra. This may have represented the fourth case of post-menopausal osteoporosis in this family of four sisters although x-rays of the fourth case have not been studied.

SUMMARY

In summary, cases of disseminated lupus erythematosis, so-called acquired porphyria, chronic macrocytic anemia with thrombocytopenic purpura, Wolff-Parkinson-White syndrome, post-menopausal osteoporosis have been observed in two or more siblings. These cases have been discovered during a relatively short period of ordinary practice in internal medicine. This suggests that a survey of the family of patients with an unusual disorder, not commonly regarded as familial, may prove a fruitful source of important and interesting pathology and may even aid in the diagnosis.

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